Case Report

Challenges in Managing Retinitis Pigmentosa: A Case Report and Review of Current Literature

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Abstract

Retinitis pigmentosa (RP) is a group of inherited retinal disorders characterized by progressive photoreceptor degeneration leading to severe vision loss. This condition poses significant challenges for early diagnosis and management because of its complex genetic basis and phenotypic variability. We report the case of a 43-year-old female diagnosed with progressive RP who presented with a familial history of the disorder. Her condition, characterized by a steady decline in visual acuity starting at age 18 years, progressed to complete loss of light perception despite regular ophthalmological assessments and interventions, including antioxidant and vitamin supplements. This case underscores the relentless progression of RP and the lack of effective treatment. This highlights the need for advanced genetic and phenotypic mapping to enhance the diagnostic accuracy and therapeutic approaches.

Keywords: Antioxidants, case report, retinal degeneration, retinitis pigmentosa

Introduction

Retinitis pigmentosa (RP) encompasses a diverse group of inherited disorders characterized by the progressive degeneration of the retina's rod and cone photoreceptors.[1] This degenerative process begins with the loss of photoreceptor cells, which subsequently leads to alterations in the retinal pigment epithelium and retinal glia, culminating in widespread degenerative changes affecting the inner neural layers of the retina, its vasculature, and the optic nerve head.[1] RP is often accompanied by various macular complications stemming from anomalies at the vitreous-retinal interface. Cystoid macular edema is the most prevalent of these complications, while vitreomacular traction syndrome is less common.[2] Individuals affected with RP typically experience nyctalopia (night blindness) and a progressive constriction of the peripheral visual field in both eyes. Ultimately, these symptoms may lead to the loss of central vision and complete blindness. The onset of the disease typically occurs in the first or second decade of life.[3] The global prevalence of RP is estimated to be approximately one in every 3500–4000 individuals, with the condition being associated with mutations in over 40 different genes.[4] Diagnostic clinical features of RP include a diminished electroretinogram (ERG) response, characteristic bone spicule pigmentation, onset of tunnel vision, and a narrowed visual field.[5] The standard therapeutic approach for RP includes Vitamin A and fish oil supplements, gene therapies, cell therapies, retinal implants, and other emerging treatments including intravitreal injections of a vascular endothelial growth factor inhibitor.[6]

Case reports are essential for understanding rare diseases, such as RP, and documenting unique presentations, associations, and treatment responses that are not typically captured in larger studies. These findings provide valuable clinical

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insights into diagnosis and management. For example, a case report of an atypical form of RP, described as unilateral pericentral RP, underscores the importance of multimodal imaging for accurate diagnosis and differentiation from the common forms.[7] Additionally, a case series on unusual RP associations revealed that common retinal diseases, such as posterior pole neovascularization, could present unusually in RP patients, highlighting the need for comprehensive patient care.[8] The utility of advanced technology in diagnosing rare retinal disorders shows how detailed case reports enhance clinical understanding and diagnostic accuracy.[9] These reports emphasize the importance of the clinical documentation of rare diseases, providing a foundation for further research and improved clinical outcomes. SCARE 2023 criteria have been followed in reporting this work.[10]

Case Report

In the case of a 43-year-old female patient, the chief complaint revolves around a gradual decline in visual acuity that initially manifested at the age of 18 years. A notable aspect of her medical history is the prevalence of RP in her mother, indicating a familial predisposition to the condition. Over the years, her vision has steadily deteriorated. She was followed up every 6 months with full ophthalmologic examinations including fundus evaluation, optical coherence tomography (OCT), and visual field testing. These assessments consistently revealed progressive peripheral field constriction, eventually resulting in tunnel vision and culminating in complete bilateral loss of light perception (LP). The right eye lost LP approximately 3 years before the left eye, with the patient first presenting with LP in the right eye at age 37, followed by LP in the left at age 40. Other causes for the visual decline, including vascular events such as optic ischemia, were clinically excluded through funduscopy and the absence of systemic risk factors.

Following thorough ophthalmological evaluation, the patient received a diagnosis of progressive RP [Figure 1]. Despite various interventions, including nutritional supplements, neuroprotective agents, and retinal stimulation therapies, there has been no observable improvement in visual acuity. However, none have resulted in observable improvement in visual acuity. Additional assessments, including electrophysiology studies [Figure 2], have indicated photoreceptor dysfunction. Macular edema and retinal detachment were further ruled out through clinical examination and OCT.

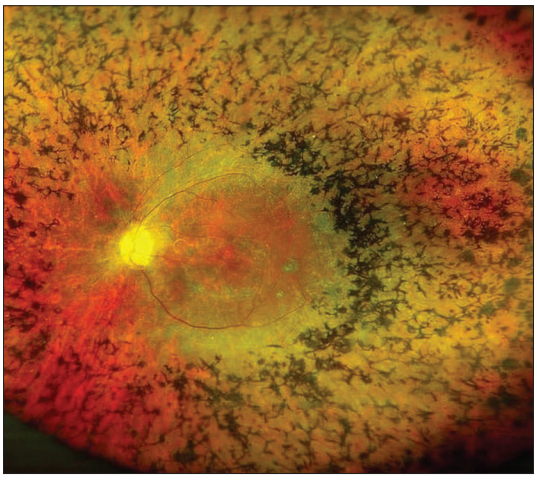

Figure 1: Fundoscopy shows hyperpigmentation in a bone-spicule configuration in the midperipheral retina, Narrowing of the retinal arterioles and disc pallor

The current management strategy involves symptomatic treatment, primarily consisting of antioxidant vitamins aimed at slowing down the disease's progression. However, it is essential to note that RP lacks a definitive cure, emphasizing the importance of preserving existing vision and preventing further deterioration. Regular follow-up appointments are scheduled to monitor disease progression, alongside counseling on lifestyle adjustments and potential referrals to low vision rehabilitation services to enhance the patient's quality of life and independence in daily activities.

Discussion

RP is a group of inherited retinal disorders characterized by the progressive degeneration of photoreceptors, leading to vision loss. As photoreceptors deteriorate, significant changes occur in retinal structure and function, which can be mapped using advanced imaging and diagnostic tools. Recent studies have utilized various mapping methods, such as electroretinography (ERG) and functional magnetic resonance imaging (fMRI), to better understand the extent and progression of retinal degeneration in patients with RP. For instance, a nonlinear ERG analysis method has shown effectiveness in distinguishing between normal and RP-affected eyes by analyzing the amplitude and implicit time changes in retinal responses to light stimuli, highlighting the potential for early diagnosis.[11] Additionally, studies on retinal remodeling using zebrafish models provide insight into molecular changes during degeneration, emphasizing the role of oxidative stress and synaptic remodeling, which complicate efforts to restore vision.[12] Furthermore, fMRI-based retinotopic mapping revealed cortical remapping in patients with RP, where visual cortex areas corresponding to degenerated retinal regions adapt by shifting their receptive fields, a phenomenon linked to the severity of visual field loss.[13] This evidence suggests that comprehensive retinal mapping is crucial for understanding RP progression and could inform future therapeutic strategies, aligning with the diagnostic methods such as retinal mapping [Figure 2] mentioned in the case presentation.

Familial predisposition plays a significant role in the manifestation of RP, and genetic testing is a crucial tool for identifying at-risk individuals and providing accurate diagnoses. Next-generation sequencing (NGS) has proven effective in detecting pathogenic mutations responsible for RP, with studies reporting a 70%–80% success rate in identifying causative genes. For instance, a study involving 107 patients with RP from India identified specific genetic

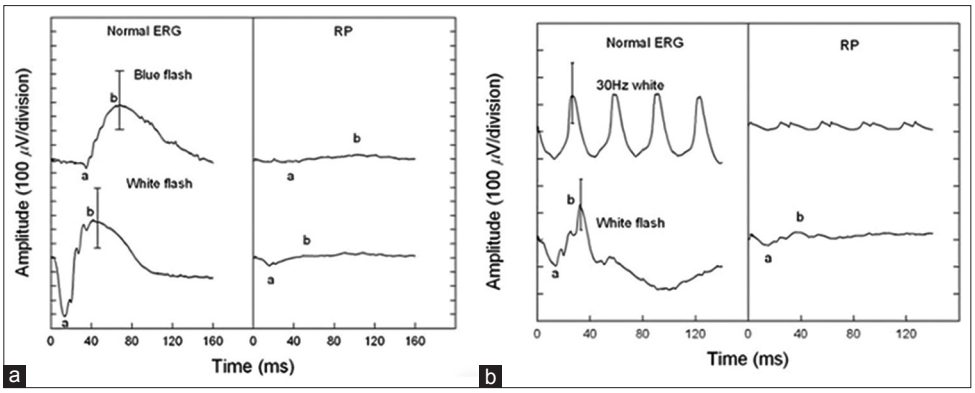

Figure 2: The electroretinogram showing attenuation in waves (a and b). ERG: Electroretinogram, RP: Retinitis pigmentosa

mutations that correlated with distinct phenotypic features such as chorioretinal atrophy in patients with CERKL and PROM1 mutations.[14] Another study on a large cohort from the University of Minnesota emphasized that NGS panels could identify genotypes consistent with clinical findings in approximately 52.8% of the patients, highlighting the importance of genetic testing for accurate diagnosis and management.[15] Furthermore, the phenotypic heterogeneity observed in the family members of patients with RP underscores the necessity for comprehensive family screening and pretest genetic counseling to effectively manage and predict disease progression.[16] These findings suggest that integrating genetic testing with familial analysis can significantly improve disease management and outcomes in patients with RP, reflecting the familial predisposition highlighted in the case presentation in which the patient's mother also had RP.

Research has explored the potential of antioxidant vitamins to slow the progression of RP. Systematic reviews suggested that, while any vitamin or mineral with antioxidant properties such as Vitamin A and E might slow the progression of other retinal diseases, such as age-related macular degeneration, evidence supporting their effectiveness in RP remains inconclusive.[17] This is in line with the case presentation, in which the patient was undergoing symptomatic treatment with antioxidant vitamins aimed at slowing disease progression. Overall, the role of antioxidants in RP management is complex and may depend heavily on the specific genetic makeup of the individual.

Recent advances in gene therapy and retinal implants have offered promising avenues for mitigating the effects of this disease. Gene therapy, particularly gene augmentation, is aimed at correcting the underlying genetic defects responsible for RP. For instance, studies of RP caused by mutations in FAM161A have demonstrated that gene augmentation can significantly restore photoreceptor function and slow degeneration in animal models, paving the way for potential human applications.[18] The Food and Drug Administration-approved voretigene neparvovec-rzyl, which targets RPE65-linked retinal dystrophy, exemplifies the success of such therapies in the clinical setting, although its applicability to other forms of RP remains challenging.[19] However, retinal implants are being explored as a complementary approach for patients with advanced stages of RP, where photoreceptor cells are severely degenerated. Although these implants do not restore natural vision, they can provide artificial vision by converting light into electrical signals that stimulate the remaining retinal cells, thus offering functional improvement in vision. The integration of gene therapy with retinal implants could potentially enhance the efficacy of treatment, offering a dual approach to combat both the genetic causes and the physiological consequences of RP.

Effective low-vision rehabilitation strategies are crucial for managing these impairments and improving the quality of life of individuals with RP. Several studies have emphasized the importance of personalized rehabilitation approaches tailored to the specific needs of each patient. For instance, Novizar *et al*. discussed the successful use of low-vision aids, such as photochromic lenses and illumination enhancements, in a pseudophakic patient with RP, leading to notable improvements in functional vision and quality of life.[20] Another study by Dewi *et al*. highlighted the importance of a comprehensive management plan, including pharmacological treatment, optical aids, and rehabilitation techniques like head tracking and scanning, to optimize the remaining vision in RP patients.[21] Jaulin focused on the role of orthoptic care in RP, emphasizing the need for early intervention to maintain mobility and daily functionality as the disease progresses.[22] These studies collectively underscore the critical role of customized low-vision rehabilitation in managing RP by offering strategies that can significantly enhance patients' independence and quality of life, reinforcing the importance of counseling on lifestyle adjustments and potential referrals to low-vision rehabilitation services mentioned in the case presentation.

The primary limitation of this case study was its reliance on observations from a single patient, which may not provide a comprehensive understanding of RP's diverse manifestations of RP or the effectiveness of various treatment strategies across different genetic backgrounds. Additionally, the case lacks longitudinal data regarding the long-term efficacy of interventions, particularly in genetically distinct populations, which could provide more insights into disease progression and response to treatment.

Conclusion

This case demonstrates the progressive nature of RP and challenges associated with its complex clinical presentation. Despite advances in diagnostic and therapeutic technologies, RP remains a debilitating condition without a cure. To effectively address RP, it is crucial to enhance genetic research on targeted gene therapies, implement comprehensive genetic screening and counseling for early diagnosis, and develop standardized guidelines for antioxidant therapy based on individual genetic profiles. Additionally, expanding low-vision rehabilitation programs tailored to patients' specific needs at various stages of vision loss will significantly improve the quality of life and functionality. These integrated approaches will better equip healthcare systems to manage RP and support affected individuals more effectively.

Authors' contributions

All authors have participated in writing the manuscript and reviewing the literature. Dr. Mohammad Zeid conceived and supervised the conduct of the study. All authors read and approved the final manuscript.

Ethical approval

Ethical approval to report this case was obtained from the Faculty of Medicine at the University of Kalamoon with serial number (836/25).

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given her consent for her images and other clinical information to be reported in the journal. The patient understands that her name and initials will not be published and due efforts will be made to conceal her identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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